



## **PRoS-FINAL-2353: Are rasopathies new monogenic predisposing conditions to the development of systemic lupus erythematosus?**

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POSTER PRESENTATION

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# PreS-FINAL-2353: Are rasopathies new monogenic predisposing conditions to the development of systemic lupus erythematosus?

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## Introduction

RASopathies (Noonan syndrome (NS) and Noonan-related syndromes) are neurodevelopmental syndromes resulting from germline mutations in genes that participate in the rat sarcoma/mitogen-activated protein kinases (RAS/MAPK) pathway (*PTPN11*, *SOS1*, *RAF*, *KRAS* or *NRAS* and *SHOC2*). Some monogenic conditions are associated with the development of systemic lupus erythematosus (SLE), and a few reports described the association of SLE, with NS.

## Objectives

We aim to search for a relationship between RASopathy and the development of SLE.

## Methods

We reported for the first time on a 13-year-old boy with NS with loose anagen hair (NSLAH) resulting from mutation in *SHOC2* who developed an autoimmune disorder which fulfilled four American College of Rheumatology (ACR) criteria for the classification of SLE (polyarthritis, pericarditis, antinuclear antibodies, anti-DNA antibodies). The case report then prompted a literature review by a systematic search for English and French articles on the subjects of RASopathies and SLE that had English abstracts in PubMed from 1966 to 2012.

## Results

We identified seven additional patients with RASopathy and SLE. The male-to-female ratio was 1:1, and age at onset of SLE ranged from 5 to 32 years. The most common features were polyarthritis (7/8 patients), auto-

immune cytopenia (4/8 patients) and pericarditis (4/8 patients) while only one patient presented with skin involvement.

## Conclusion

The association of two rare diseases in eight patients suggests that RASopathies may be associated with the development of SLE, which is characterized by a higher male-to-female ratio, a lower rate of skin involvement and a higher rate of pericarditis than "classic" SLE.

## Disclosure of interest

None declared.

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